Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Sickle Cell Disease and Other Hemoglobinopathies

Volume 15, No. 2 | Quarter 2 | May 2005

INTRODUCTION

On April 4, 2005, we distributed to all active participants the Quarter 2 proficiency testing (PT) panel consisting of five dried-blood-spot (DBS) specimens for sickle cell disease and other hemoglobinopathies. A total of 68 PT panels were mailed by overnight FedEx mail. The packages went to 59 domestic laboratories and 9 foreign laboratories. The specimen panel consisted of five DBS specimens prepared from umbilical cord blood. This PT report is a compilation of all data reports for hemoglobinopathy testing received from participants by the designated deadline date. We distribute this quarterly report to all participants, state laboratory directors, and to program colleagues by request. We received data reports from 61 newborn screening laboratories. There were 7 laboratories that did not report this quarter.
We requested that participants assay all survey specimens by the analytic schemes they routinely use and report for each specimen the presumptive phenotype, the presumptive clinical assessment, and any other clinical classifications that they deem consistent with their analytic results and program operations. ❖

PARTICIPANTS' RESULTS

The certification report listing hemoglobins (Hbs) by phenotype and their presumptive clinical assessments appears on page 2.

The frequency distribution of reported phenotypes and presumptive clinical assessments appears on page 3.

The individual data verification for each laboratory with evaluation comments appears on page 4.

This quarter there were no misclassifications. "

The NSQAP will ship next quarter's PT specimens on July 11, 2005.❖

SPOTLIGHT
Meetings

The upcoming meeting of the Sickle Cell Disease Advisory Committee will be held on Monday, June 6, 2005, at 8:30 a.m., 6701 Rockledge Drive, Rockledge II, Conference Room 9112. Dr. Harry Hannon will attend the meeting as the NSQAP representative.

Articles of Interest

"Systematic follow-up and case management of the abnormal newborn screen can improve acceptance of genetic counseling for sickle cell or other hemoglobinopathy trait", Beth Kladny, Ms, CGC, Elizabeth A. Gettig, MS, CGC, and Lakshmanan Krishnamuriti, MD Genet Med February 2005:7(2):139-142.❖

ACKNOWLEDGMENTS

The specimens for this survey were prepared from umbilical cord blood samples supplied by Alabama State Public Health Laboratory.❖

Do you know what countries participate in the NSQAP hemoglobinopathies program?

United States
Brazil
Chile
South Korea
Spain
United Kingdom

CDC/APHL

This program is cosponsored by the Centers for Disease Control and Prevention (CDC) and the Association of Public Health Laboratories (APHL).

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CENTERS FOR DISEASE CONTROL AND PREVENTION

Newborn Screening Quality Assurance Program
Sickle Cell Disease and Other Hemoglobinopathies

Specimen Certification Report

Year: 2005 Quarter: 2

Presumptive Clinical Phenotypes

	Specimen 2531	Specimen 2532	Specimen 2533	Specimen 2534	Specimen 2535
Expected Presumptive Phenotype	FAC	FS	FSC	FAS	FA
Accepted Presumptive Phenotypes	FCA, AFC		FCS	AFS	

Presumptive Clinical Assessments

	Specimen 2531	Specimen 2532	Specimen 2533	Specimen 2534	Specimen 2535
Expected Presumptive Clinical Assessment	03	04	05	02	01
Accepted Presumptive Clinical Assessments	15, 21	13, 21	21	15, 21	21

01 Normal--no abnormal Hb found
02 Hemoglobin S carrier
03 Hemoglobin C carrier
04 Hemoglobin S, S disease (Sickle cell anemia)
05 Hemoglobin S, C disease
06 Hemoglobin S, D disease
07 Hemoglobin S, O disease
08 Hemoglobin D carrier
09 Hemoglobin E carrier
10 Hemoglobin G carrier
11 Hemoglobin O carrier
12 Hemoglobin S, E disease
13 Hemoglobin S Beta-thalassemia
14 Hemoglobin E Beta-thalassemia
15 Hemoglobin C Beta-thalassemia
16 Alpha-thalassemia (Bart's Hb)
17 Transfused infant
18 Hemoglobin E, E disease
19 Combination one or more Hbs
20 Assessment is not listed
21 Unsatisfactory specimen
22 Unidentified Variant
23 Hemoglobin E Alpha-thalassemia
24 Hemoglobin D Beta-thalassemia
(NE) Specimen not evaluated

Newborn Screening Quality Assurance Program
Sickle Cell Disease and Other Hemoglobinopathies
Frequency Distributions

Year: 2005 Quarter: 2

Phenotypes

Specimen Number	Hemoglobin Phenotypes	Frequency Distributions
2531	AFC	1
	FAC	55
	FCA	5
2532	FS	61
2533	FCS	8
	FSC	53
2534	AFS	1
	FAS	60
2535	FA	61

Clinical Assessments

Specimen Number	Presumptive Assessments	Frequency Distributions
2531	03 HbC carrier	61
2532	04 Sickle cell anemia	61
2533	05 Sickle-Hb C disease	61
2534	02 HbS carrier	61
2535	01 Normal	61